Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

Genetics and Human Traits

Reprinted from Genetics Home Reference (http://ghr.nlm.nih.gov/)

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services

Published June 29, 2015

Chapter 5

Genetics and Human Traits

Table of Contents

Are fingerprints determined by genetics?

Each person's fingerprints are unique, which is why they have long been used as a way to identify individuals. Surprisingly little is known about the factors that influence a person's fingerprint patterns. Like many other complex (http://ghr.nlm.nih.gov/handbook/mutationsanddisorders/complexdisorders) traits, studies suggest that both genetic and environmental factors play a role.

A person's fingerprints are based on the patterns of skin ridges (called dermatoglyphs) on the pads of the fingers. These ridges are also present on the toes, the palms of the hands, and the soles of the feet. Although the basic whorl, arch, and loop patterns may be similar, the details of the patterns are specific to each individual.

Dermatoglyphs develop before birth and remain the same throughout life. The ridges begin to develop during the third month of fetal development, and they are fully formed by the sixth month. The function of these ridges is not entirely clear, but they likely increase sensitivity to touch.

The basic size, shape, and spacing of dermatoglyphs appear to be influenced by genetic factors. Studies suggest that multiple genes are involved, so the inheritance pattern is not straightforward. Genes that control the development of the various layers of skin, as well as the muscles, fat, and blood vessels underneath the skin, may all play a role in determining the pattern of ridges. The finer details of the patterns of skin ridges are influenced by other factors during fetal development, including the environment inside the womb. These developmental factors cause each person's dermatoglyphs to be different from everyone else's. Even identical twins, who have the same DNA, have different fingerprints.

Few genes involved in dermatoglyph formation have been identified. Rare diseases characterized by abnormal or absent dermatoglyphs provide some clues as to their genetic basis. For example, a condition known as adermatoglyphia is characterized by an absence of dermatoglyphs, sometimes with other abnormalities of the skin. Adermatoglyphia is caused by mutations in a gene called *SMARCAD1*. Although this gene is clearly important for the formation of dermatoglyphs, its role in their development is unclear.

To find out more about the influence of genetics on the formation of fingerprints:

The UCSB Science Line from the University of California, Santa Barbara provides information about how fingerprints are formed (http://scienceline.ucsb.edu/getkey.php?key=2650).

The Mad Sci Network offers many Q&As related to fingerprints (http://www.madsci.org/FAQs/body/fingerprints.html), including the genetics and

development of dermatoglyphs. The questions were asked by students and answered by scientists.

This article from Science Magazine discusses the case of a woman with adermatoglyphia (http://news.sciencemag.org/plants-animals/2011/08/mystery-missing-fingerprints).

OMIM.org provides more detailed genetic information about dermatoglyphs (http://omim.org/entry/125590) and adermatoglyphia (http://omim.org/entry/136000).

Scientific journal articles for further reading:

Burger B, Fuchs D, Sprecher E, Itin P. The immigration delay disease: adermatoglyphia-inherited absence of epidermal ridges. J Am Acad Dermatol. 2011 May;64(5):974-80. doi: 10.1016/j.jaad.2009.11.013. Epub 2010 Jul 8.PMID 20619487 (http://www.ncbi.nlm.nih.gov/pubmed/20619487).

Nousbeck J, Burger B, Fuchs-Telem D, Pavlovsky M, Fenig S, Sarig O, Itin P, Sprecher E. A mutation in a skin-specific isoform of *SMARCAD1* causes autosomal-dominant adermatoglyphia. Am J Hum Genet. 2011 Aug 12;89(2):302-7. doi: 10.1016/j.ajhg.2011.07.004. Epub 2011 Aug 4. PMID 21820097 (http://www.ncbi.nlm.nih.gov/pubmed/21820097). Free full-text available from PubMed Central: PMC3155166 (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3155166/).

Warman PH, Ennos A.R. Fingerprints are unlikely to increase the friction of primate fingerpads. J Exp Biol. 2009 Jul;212(Pt 13):2016-22. doi: 10.1242/jeb.028977. PMID 19525427 (http://www.ncbi.nlm.nih.gov/pubmed/19525427).

Is eye color determined by genetics?

A person’s eye color results from pigmentation of a structure called the iris, which surrounds the small black hole in the center of the eye (the pupil) and helps control how much light can enter the eye. The color of the iris ranges on a continuum from very light blue to dark brown. Most of the time eye color is categorized as blue, green/hazel, or brown. Brown is the most frequent eye color worldwide. Lighter eye colors, such as blue and green, are found almost exclusively among people of European ancestry.

Eye color is determined by variations in a person’s genes. Most of the genes associated with eye color are involved in the production, transport, or storage of a pigment called melanin. Eye color is directly related to the amount and quality of melanin in the front layers of the iris. People with brown eyes have a large amount of melanin in the iris, while people with blue eyes have much less of this pigment.

A particular region on chromosome 15 plays a major role in eye color. Within this region, there are two genes located very close together: *OCA2* and *HERC2*. The protein produced from the *OCA2* gene, known as the P protein, is involved in the maturation of melanosomes, which are cellular structures that produce and store melanin. The P protein therefore plays a crucial role in the amount and quality of melanin that is present in the iris. Several common variations (polymorphisms) in the *OCA2* gene reduce the amount of functional P protein that is produced. Less P protein means that less melanin is present in the iris, leading to blue eyes instead of brown in people with a polymorphism in this gene.

A region of the nearby *HERC2* gene known as intron 86 contains a segment of DNA that controls the activity (expression) of the *OCA2* gene, turning it on or off as needed. At least one polymorphism in this area of the *HERC2* gene has been shown to reduce the expression of *OCA2*, which leads to less melanin in the iris and lighter-colored eyes.

Several other genes play smaller roles in determining eye color. Some of these genes are also involved in skin and hair coloring. Genes with reported roles in eye color include *ASIP*, *IRF4*, *SLC24A4*, *SLC24A5*, *SLC45A2*, *TPCN2*, *TYR*, and *TYRP1*. The effects of these genes likely combine with those of *OCA2* and *HERC2* to produce a continuum of eye colors in different people.

Researchers used to think that eye color was determined by a single gene and followed a simple inheritance pattern in which brown eyes were dominant to blue eyes. Under this model, it was believed that parents who both had blue eyes could not have a child with brown eyes. However, later studies showed that this model was too simplistic. Although it is uncommon, parents with blue eyes can have children with brown eyes. The inheritance of eye color is more complex than

originally suspected because multiple genes are involved. While a child's eye color can often be predicted by the eye colors of his or her parents and other relatives, genetic variations sometimes produce unexpected results.

Several disorders that affect eye color have been described. Ocular albinism is characterized by severely reduced pigmentation of the iris, which causes very light-colored eyes and significant problems with vision. Another condition called oculocutaneous albinism affects the pigmentation of the skin and hair in addition to the eyes. Affected individuals tend to have very light-colored irises, fair skin, and white or light-colored hair. Both ocular albinism and oculocutaneous albinism result from mutations in genes involved in the production and storage of melanin. Another condition called heterochromia is characterized by different-colored eyes in the same individual. Heterochromia can be caused by genetic changes or by a problem during eye development, or it can be acquired as a result of a disease or injury to the eye.

To learn more about the genetics of eye color:

John H. McDonald at the University of Delaware discusses the myth that eye color is determined by a single gene (http://udel.edu/~mcdonald/mytheyecolor.html).

The Tech Museum of Innovation at Stanford University provides a Q&A explaining how brown-eyed parents can have blue-eyed children (http://genetics.thetech.org/ask-a-geneticist/brown-eyed-parents-blue-eyed-kids).

The University of Kansas Medical Center offers links to additional resources (https://www.kumc.edu/gec/support/eyecolor.html) about the genetics of eye and hair color.

More detailed information about ocular albinism (http://omim.org/entry/300500) and oculocutaneous albinism (http://omim.org/entry/203100), as well as the genetics of eye, hair, and skin color variation (http://omim.org/entry/227220), is available from OMIM.org.

A brief description of heterochromia (http://www.nlm.nih.gov/medlineplus/ency/article/003319.htm) is available from MedlinePlus.

Scientific journal articles for further reading:

Sturm RA, Duffy DL, Zhao ZZ, Leite FP, Stark MS, Hayward NK, Martin NG, Montgomery GW. A single SNP in an evolutionary conserved region within intron 86 of the *HERC2* gene determines human blue-brown eye color. Am J Hum Genet. 2008 Feb;82(2):424-31. doi: 10.1016/j.ajhg.2007.11.005. Epub 2008 Jan 24. PMID 18252222 (http://www.ncbi.nlm.nih.gov/pubmed/18252222). Free full-text available from PubMed Central: PMC2427173 (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2427173/).

Sturm RA, Larsson M. Genetics of human iris colour and patterns. Pigment Cell Melanoma Res. 2009 Oct;22(5):544-62. doi: 10.1111/j.1755-148X.2009.00606.x. Epub 2009 Jul 8. Review. PMID: 19619260 (http://www.ncbi.nlm.nih.gov/pubmed/19619260).

White D, Rabago-Smith M. Genotype-phenotype associations and human eye color. J Hum Genet. 2011 Jan;56(1):5-7. doi: 10.1038/jhg.2010.126. Epub 2010 Oct 14. Review. PMID: 20944644 (http://www.ncbi.nlm.nih.gov/pubmed/20944644)

Is intelligence determined by genetics?

Like most aspects of human behavior and cognition, intelligence is a complex (http://ghr.nlm.nih.gov/handbook/mutationsanddisorders/complexdisorders) trait that is influenced by both genetic and environmental factors.

Intelligence is challenging to study, in part because it can be defined and measured in different ways. Most definitions of intelligence include the ability to learn from experiences and adapt to changing environments. Elements of intelligence include the ability to reason, plan, solve problems, think abstractly, and understand complex ideas. Many studies rely on a measure of intelligence called the intelligence quotient (IQ).

Researchers have conducted many studies to look for genes that influence intelligence. Many of these studies have focused on similarities and differences in IQ within families, particularly looking at adopted children and twins. These studies suggest that genetic factors underlie about 50 percent of the difference in intelligence among individuals. Other studies have examined variations across the entire genomes of many people (an approach called genome-wide association studies (http://ghr.nlm.nih.gov/handbook/genomicresearch/gwastudies) or GWAS) to determine whether any specific areas of the genome are associated with IQ. These studies have not conclusively identified any genes that underlie differences in intelligence. It is likely that a large number of genes are involved, each of which makes only a small contribution to a person's intelligence.

Intelligence is also strongly influenced by the environment. Factors related to a child's home environment and parenting, education and availability of learning resources, and nutrition, among others, all contribute to intelligence. A person's environment and genes influence each other, and it can be challenging to tease apart the effects of the environment from those of genetics. For example, if a child's IQ is similar to that of his or her parents, is that similarity due to genetic factors passed down from parent to child, to shared environmental factors, or (most likely) to a combination of both? It is clear that both environmental and genetic factors play a part in determining intelligence.

To find out more about the influence of genetics on intelligence:

This news release from the journal Nature explains why it is so difficult to identify genes associated with IQ: "'Smart genes' prove elusive" (http://www.nature.com/news/smart-genes-prove-elusive-1.15858) (September 8, 2014)

The Tech Museum of Innovation at Stanford University provides a Q&A about the influence of genes and environment on IQ (http://genetics.thetech.org/ask-a-geneticist/intelligence-and-genetics).

The Cold Spring Harbor Laboratory offers an interactive tool called Genes to Cognition (http://www.g2conline.org/) that provides information about many aspects of the genetics of neuroscience.

Scientific journal articles for further reading:

Deary IJ. Intelligence. Curr Biol. 2013 Aug 19;23(16):R673-6. doi: 10.1016/j.cub.2013.07.021. PMID: 23968918 (http://www.ncbi.nlm.nih.gov/pubmed/23968918). Free full-text available from the publisher: http://www.sciencedirect.com/science/article/pii/S0960982213008440

Deary IJ, Johnson W, Houlihan LM. Genetic foundations of human intelligence. Hum Genet. 2009 Jul;126(1):215-32. doi: 10.1007/s00439-009-0655-4. Epub 2009 Mar 18. Review. PMID: 19294424 (http://www.ncbi.nlm.nih.gov/pubmed/19294424).

Plomin R, Deary IJ. Genetics and intelligence differences: five special findings. Mol Psychiatry. 2015 Feb;20(1):98-108. doi: 10.1038/mp.2014.105. Epub 2014 Sep 16. Review. PMID: 25224258 (http://www.ncbi.nlm.nih.gov/pubmed/25224258). Free full-text available from PubMed Central: PMC4270739 (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4270739/).

Sternberg RJ. Intelligence. Dialogues Clin Neurosci. 2012 Mar;14(1):19-27. Review. PMID: 22577301 (http://www.ncbi.nlm.nih.gov/pubmed/22577301). Free full-text available from PubMed Central: PMC3341646 (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3341646/)

Genetics Home Reference
Your Guide to Understanding Genetic Conditions

http://ghr.nlm.nih.gov/

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services

Published on June 29, 2015